Sturge-Weber syndrome

Description

Sturge-Weber syndrome is a condition that affects the development of certain blood vessels, causing abnormalities in the brain, skin, and eyes from birth. Sturge-Weber syndrome has three major features: a red or pink birthmark called a port-wine birthmark, a brain abnormality called a leptomeningeal angioma, and increased pressure in the eye (glaucoma). These features can vary in severity and not all individuals with Sturge-Weber syndrome have all three features.

Most people with Sturge-Weber syndrome are born with a port-wine birthmark. This type of birthmark is caused by enlargement (dilatation) of small blood vessels (capillaries) near the surface of the skin. Port-wine birthmarks are typically initially flat and can vary in color from pale pink to deep purple. In people with Sturge-Weber syndrome, the port-wine birthmark is most often on the face, typically on the forehead, temple, or eyelid. The port-wine birthmark is usually only on one side of the face but can be on both sides. Over time, the skin within the port-wine birthmark can darken and thicken.

In Sturge-Weber syndrome, there is usually abnormal formation and growth of blood vessels within the two thin layers of tissue that cover the brain and spinal cord. This abnormality, which is called leptomeningeal angioma, can affect one or both sides of the brain and impair blood flow in the brain and lead to loss of brain tissue (atrophy) and deposits of calcium (calcification) in the brain below the angioma. The decrease in blood flow caused by leptomeningeal angiomas can cause stroke-like episodes in people with Sturge-Weber syndrome. These episodes often involve temporary muscle weakness on one side of the body (hemiparesis), vision abnormalities, seizures, and migraine headaches. In affected individuals, these episodes usually begin by age 2. The seizures usually involve only one side of the brain (focal seizures), during which the port-wine birthmark may darken and individuals may lose consciousness. People with Sturge-Weber syndrome have varying levels of cognitive function, from normal intelligence to intellectual disability. Some individuals have learning disabilities with problems focusing similar to attention-deficit/hyperactivity disorder (ADHD).

In individuals with Sturge-Weber syndrome, glaucoma typically develops either in infancy or early adulthood and can cause vision impairment. In some affected infants, the pressure can become so great that the eyeballs appear enlarged and bulging (buphthalmos). Individuals with Sturge-Weber syndrome can have tangles of abnormal blood vessels (hemangiomas) in various parts of the eye. When these abnormal blood

vessels develop in the network of blood vessels at the back of the eye (choroid), it is called a diffuse choroidal hemangioma and occurs in about one-third of individuals with Sturge-Weber syndrome. A diffuse choroidal hemangioma can cause vision loss. When present, the eye abnormalities typically occur on the same side of the head as the port-wine birthmark.

Frequency

Sturge-Weber syndrome is estimated to affect 1 in 20,000 to 50,000 individuals.

Causes

Sturge-Weber syndrome is caused by a mutation in the *GNAQ* gene. This gene provides instructions for making a protein called guanine nucleotide-binding protein G(q) subunit alpha (Gαq). The Gαq protein is part of a group of proteins (complex) that regulates signaling pathways to help control the development and function of blood vessels.

The *GNAQ* gene mutation that causes Sturge-Weber syndrome results in the production of a protein with impaired function. As a result, the altered Gαq protein cannot play its part in regulating signaling pathways, resulting in abnormally increased signaling. The enhanced signaling likely disrupts the regulation of blood vessel development, causing abnormal and excessive formation of vessels before birth in people with Sturge-Weber syndrome.

Learn more about the gene associated with Sturge-Weber syndrome

- GNAQ

Inheritance

Sturge-Weber syndrome is not inherited. The mutation that causes this disorder is somatic, which means it occurs after conception. In Sturge-Weber syndrome, the mutation is thought to occur in a cell during early development before birth. As that cell continues to grow and divide, the cells derived from it, specifically certain cells in the brain, eyes, and skin that are involved in blood vessel formation, also have the mutation, while the body's other cells do not. This situation is called mosaicism. The mosaic nature of the mutations helps to explain why the abnormal blood vessel growth occurs in some parts of the body but not in others.

Other Names for This Condition

- Angiomatosis aculoorbital-thalamic syndrome
- Encephalofacial hemangiomatosis
- Encephalofacial hemangiomatosis syndrome
- Meningo-oculo-facial angiomatosis

- Meningofacial angiomatosis-cerebral calcification syndrome
- Neuroretinoangiomatosis
- Phakomatosis, Sturge-Weber
- Sturge-Weber-Dimitri syndrome
- Sturge-Weber-Krabbe syndrome
- SWS

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Sturge-Weber syndrome (https://www.ncbi.nlm.nih.gov/gtr/conditions/C0038505/)

Genetic and Rare Diseases Information Center

- Sturge-Weber syndrome (https://rarediseases.info.nih.gov/diseases/7706/index)

Patient Support and Advocacy Resources

- National Organization for Rare Disorders (NORD) (https://rarediseases.org/)

Clinical Trials

- ClinicalTrials.gov (https://clinicaltrials.gov/search?cond=%22Sturge-Weber syndrome%22)

Catalog of Genes and Diseases from OMIM

- STURGE-WEBER SYNDROME; SWS (https://omim.org/entry/185300)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28Sturge-Weber+Syndrome%5BMAJR%5D%29+AND+%28Sturge-Weber+syndrome%5BTI%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1440+days%22%5Bdp%5D)

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